Familial caudal dysgenesis

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Familial caudal dysgenesis. ORPHA:1768

Familial caudal dysgenesis is a rare, genetic, developmental defect during embryogenesis disorder characterized by varying degrees of caudal dysgenesis, ranging from a single umbilical artery or imperforate anus to full sirenomelia, in several members of the same family. Phenotype includes lumbosacral agenesis, anal atresia or ectopia, genitourinary abnormalities, components of VATER or VACTERL association, and facial dysmorphism (flat facies, abnormal ears, bilateral epicanthic folds, depressed nasal bridge, micrognathia). Additional features reported include cardiovascular (e.g. endocardial cushion defect, hypoplasia of pulmonary artery) and skeletal (kyphosis, hemipelvis) anomalies.